Small patient, big challenges: Successful perioperative management of Mucopolysaccharidosis Type II with hypertrophic cardiomyopathy: A case report

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Abstract

A high anaesthetic risk in Mucopolysaccharidosis (MPS) is mainly because of anticipated difficult airway and presence of comorbidities. This underlines the critical role of an appropriate anaesthetic plan. We present a case of 11month old male baby weighing 7.6 kg diagnosed with MPS II with non-obstructive hypertrophic cardiomyopathy with dilated LA/LV with MVP with MR and mild AR posted for elective bilateral herniotomy. Baby was given syrup Triclofos 600mg 90 minutes prior induction and shifted inside operating room. All standard monitors were attached. Iv line secured with 1- 2% sevoflurane along with O2 and induced with inj. Propofol 20mg, inj. Fentanyl 20mcgs. After check ventilation, Igel 1.5 was secured and fixed after checking bilateral equal air entry and was kept on spontaneous ventilation. Further, maintained with O2 and N2O along with 1-2% sevoflurane. Baby turned into lateral position and caudal anaesthesia was administered with inj. L-Bupivacaine 0.25% 8ml in graded doses. As surgeon felt hernial sacs were big and abdominal contents were protruding into scrotum and for better surgical comfort, inj. Atracurium 3mg was given and put on controlled ventilation. There were 2 episodes of hypotension intraoperatively and were maintained with 1mg inj. Mephentermin. Rest intra-op was uneventful. Baby was extubated in deeper plane with inj. Glycopyrrolate 0.1mg + inj. Neostigmine 0.4mg. Post extubation was uneventful. The coarse facial features have posed little issue with mask ventilation. Hypertrophic biventricular cardiomyopathy might cause stress response leading to hemodynamic instability during laryngoscopy, which was overcome by securing LMA and deeper extubation.

Keywords: mucopolysacharidosis, hypertrophic cardiomyopathy, mitral valve prolapse, laryngeal mask airway

Introduction

Mucopolysaccharidoses (MPS) are a group of rare lysosomal storage disorders characterized by defective degradation of glycosaminoglycans (GAGs), resulting in their accumulation in various tissues. Among the seven types and numerous subtypes of MPS, Hunter syndrome (MPS II) is an X-linked recessive disorder with an estimated incidence of 1 in 100,000 to 170,000 live births, predominantly affecting males. It results from a deficiency of the enzyme iduronate-2-sulfatase, leading to progressive multisystemic involvement[1].

The clinical manifestations of MPS II are diverse and affect multiple organ systems, including the respiratory, cardiovascular, skeletal, and central nervous systems. Patients typically present with coarse facial features, short stature, skeletal deformities, joint stiffness, and varying degrees of intellectual disability. Cardiovascular involvement is common, with approximately 60-90% of patients developing cardiac abnormalities, including valvular disease, cardiomyopathy, and coronary artery disease[2].

The co-occurrence of MPS II with hypertrophic cardiomyopathy (HCM) presents unique challenges for anaesthetic management during surgical procedures. HCM is characterized by inappropriate myocardial hypertrophy that can lead to dynamic left ventricular outflow tract obstruction, diastolic dysfunction, mitral regurgitation, and arrhythmias. The anaesthetic implications of this combination are significant, requiring careful consideration of drug selection, hemodynamic monitoring, and airway management.

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Current anesthetic literature presents a significant knowledge gap regarding the perioperative management of pediatric patients presenting with the dual pathology of mucopolysaccharidosis type II (MPS II) and hypertrophic cardiomyopathy. While isolated case reports exist for each condition independently, no previous studies have comprehensively addressed the unique anesthetic challenges posed by this rare clinical combination. This case report presents the first detailed documentation of successful anesthetic management in an 11-month-old infant with concurrent MPS II and non-obstructive hypertrophic cardiomyopathy undergoing bilateral herniotomy. Our findings provide novel insights into the complex interplay between these pathophysiologic processes and establish evidence-based perioperative protocols. The comprehensive analysis of preoperative optimization strategies, intraoperative monitoring considerations, and postoperative management protocols presented herein fills a critical void in pediatric anesthesia literature and provides essential guidance for clinicians managing this rare but challenging patient population. This report represents the first systematic approach to understanding the synergistic anesthetic implications of these co-existing conditions in the pediatric population.

Case report

- We present a case of 11month old male baby, born out of 2nd degree consanguineous marriage, weighing 7.6 kilograms (grade I PEM) (0 to -2SD), height of 68 centimetres (-3SD) diagnosed with MPS II with non-obstructive hypertrophic cardiomyopathy with dilated left atrium/left ventricle with valvular lesions posted for elective bilateral herniotomy.
- Patient was diagnosed with MPS and Cardiomyopathy at 8th month of life. On pre-anaesthetic evaluation, a detailed head to toe examination revealed - coarse facial features, hypertelorism, frontal bossing, depressed nasal bridge, upturned nose, low set ears with right pre auricular tags, short neck, pectus carinatum and pectus excavatum, mongolian spots over lower back. Baby had a repeated hospitalization for recurrent respiratory tract infections in the past. Other routine blood investigations were within normal limits.
- Echocardiography done at 8th month of life revealed - hypertrophic cardiomyopathy with dilated left atrium/left ventricle with mild left ventricular dysfunction with Mitral Valve Prolapse with grade II Mitral Regurgitation and mild Aortic Regurgitation. A repeat check echocardiography was done prior surgery, revealed the same. Ultrasonography abdomen showed – a ~1.5 x 1.2 mm haemangioma in right lobe of liver. High risk consent was obtained.

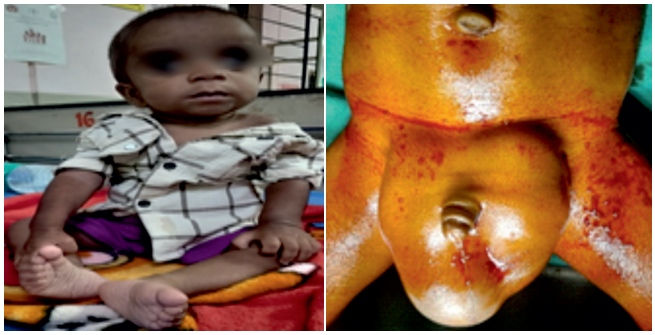

Figure 1: Characteristic features and inguinal swelling

Anaesthetic management

On the day of surgery, baby was given, syrup. Triclofos 600mg per orally, 90 minutes prior the surgery. After 60 minutes, when baby was lying calmly, gastric ultrasound was performed to confirm empty stomach. Then baby was shifted to the operating theatre.

- All standard monitors - ECG, Pulse oximeter, NIBP, nasal etCO_2 were connected.
- IV line of 24G was secured using 2% Sevoflurane along with Oxygen. IVF started with Ringers lactate at a calculated dosage.
- Induced with inj. Propofol 20mg IV, inj. Fentanyl 20mcgs IV. After check ventilation, I gel 1.5 was secured and fixed after confirming bilateral equal air entry. Baby was kept on spontaneous ventilation. Baby turned into left lateral position, under strict aseptic precaution, parts painted and draped. Caudal anaesthesia administered with 0.25% inj. L-Bupivacaine 8 ml in graded doses. Adequate block was achieved. Hemodynamics were stable.

Intraoperative Management

Figure 2: showing intraoperative management

Anaesthesia was maintained with Oxygen + Nitrous oxide mixture (50:50), Sevoflurane (1-2 vol%). As surgeon felt hernial sacs were big and abdominal contents were protruding into scrotum, for a better surgical comfort, we decided to give muscle relaxation using inj. Atracurium 3mg IV and was put on controlled ventilation using Jackson Rees circuit. Hemodynamics were monitored. Intraoperatively, there were 2 episodes of hypotension, which was managed with inj. Mephentermine 1mg IV. Rest intra-op was uneventful. Neuromuscular blockade was reversed with inj. Glycopyrrolate 0.1mg IV + inj. Neostigmine 0.4mg IV. Baby was extubated in a deeper plane and shifted to the PICU. Postoperative period was uneventful. Patient was hemodynamically stable in PICU and was discharged on 3rd post operative day.

Discussion

MPS II (50%) is the most common among all other MPS I (13.7%), MPS III (7.9%), MPS IV (24%), MPS VI (2.6%). A high percentage of MPS patients require surgery at some point of life. Most common surgical interventions: adenotonsillectomy, tympanostomy, inguinal repair, and release of carpel tunnel.

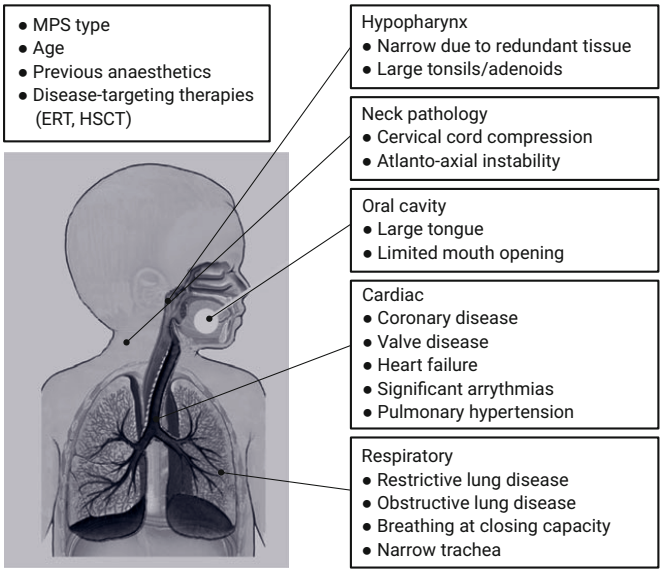

Figure 3: Showing overview of anaesthetic risk factors in patients with MPS. *ERT* enzyme replacement therapy, *HSCT* hematopoietic stem-cell transplantation[3].

Common features like respiratory limitations, cardiovascular diseases, skeletal dysfunction, facial dysmorphism, neurological symptoms, ocular disorders pose a high anaesthetic risk. Thoracic cage abnormalities predispose to the development of respiratory restrictive diseases, which often lead to the development of OSAS, pulmonary hypertension, cor pulmonale and eventually respiratory failure.

This case highlights the anaesthetic challenges of managing a paediatric patient with Mucopolysaccharidosis Type II (Hunter syndrome) with associated cardiac complications. The anaesthetic management was complex due to multiple considerations including the patient's age (11 months), potential airway difficulties, cardiac abnormalities, and the need for adequate surgical anaesthesia with muscle relaxation as they might have larger surgical defects.

Airway management considerations

The described facial features (coarse facies, hypertelorism, frontal bossing, depressed nasal bridge) are classic findings in MPS disorders which not only predict difficult airway but also difficult mask ventilation[4]. Our team appropriately opted for an I-gel supraglottic airway device initially while maintaining spontaneous ventilation, which is a prudent approach in patients with anticipated difficult airways. This strategy allows for oxygenation and ventilation while minimizing airway manipulation.

Walker et al. noted that patients with MPS often have a high incidence of difficult intubation, with one series reporting difficult laryngoscopy in 54% of MPS patients[3]. The glycosaminoglycan deposition in soft tissues often leads to tongue enlargement, adeno-tonsillar hypertrophy, and thickened supraglottic tissues. Though not needed in this case, video laryngoscopy should be considered for definitive airway management in MPS patients [5].

Cardiac implications for anaesthesia

The significant cardiac involvement in this patient (hypertrophic cardiomyopathy with dilated chambers, valvular lesions) posed additional anaesthetic challenges. Frawley et al. found that cardiovascular abnormalities are present in up to 90% of MPS II patients, requiring careful hemodynamic management [6]. The team's vigilant monitoring and prompt treatment of hypotensive episodes with mephentermine was appropriate given the patient's cardiac status.

Hemodynamic goals in hypertrophic cardiomyopathy include maintaining adequate preload, avoiding excessive afterload reduction, maintaining sinus rhythm, and avoiding tachycardia [7]. The combination of a balanced anaesthetic technique (sevoflurane, fentanyl) with regional anaesthesia (caudal block) helped maintain stable hemodynamics while providing adequate analgesia.

Regional anaesthesia considerations

The use of caudal anaesthesia with L-bupivacaine was a sound decision that likely contributed to stable hemodynamics and provided excellent postoperative analgesia. However, when administering neuraxial

blocks in MPS patients, careful consideration of potential spinal abnormalities is warranted. Solanki et al. reported that spinal cord compression and atlantoaxial instability can occur in MPS patients, particularly in types IV and VI, but can also present in type II[8].

Muscle relaxation decision

The intraoperative decision to administer atracurium and convert to controlled ventilation demonstrates flexibility in anaesthetic management based on surgical needs. However, the decision to use deep sedation must be made based on the respiratory conditions of the individual patient; some of them are eligible for deep sedation with native airways[9] while, for others, LMA in spontaneous ventilation or general anesthesia with endotracheal intubation is preferable[10]. The dosing appears appropriate for a short procedure, and reversal was adequately performed prior to extubation.

Perioperative management

The preoperative administration of Triclofos for anxiolysis followed by gastric ultrasound to confirm gastric emptying demonstrates thoughtful planning, given that MPS patients may have delayed gastric emptying[1]. The decision to extubate in a deeper plane (as we were able to mask ventilate preoperatively) is controversial but was likely made to minimize the risk of laryngospasm during emergence, which is higher in patients with abnormal airway anatomy.

Moretto et al. emphasized the importance of close postoperative monitoring in MPS patients due to the risk of upper airway obstruction [10], and the decision to admit the patient to PICU for observation was appropriate.

Conclusion

This case exemplifies successful anaesthetic management of a challenging paediatric patient with MPS II and cardiac complications. The management approach followed key principles recommended for MPS patients: thorough preoperative assessment, careful airway management, vigilant hemodynamic monitoring, and appropriate use of regional techniques. The uneventful recovery and discharge on postoperative day 3 reflect effective perioperative care.

For future anaesthetic management of this patient, preparation for progressive airway and cardiac complications should be anticipated as the disease advances, and coordination with a multidisciplinary team including cardiologists and intensivists would be beneficial.

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